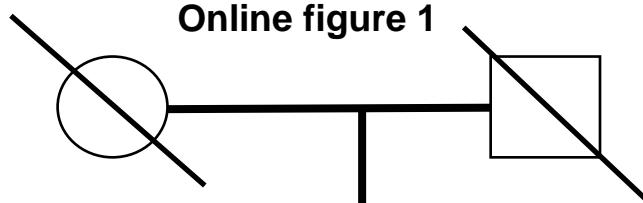


LEPR T699M

Online figure 1

Family F1

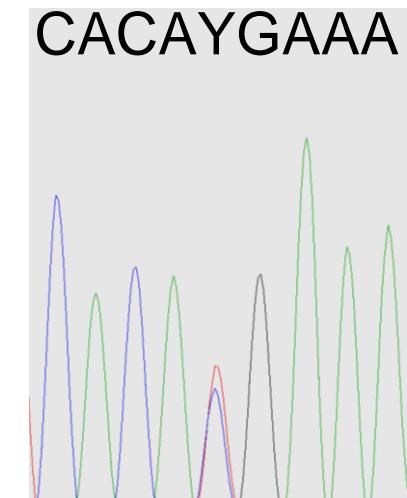
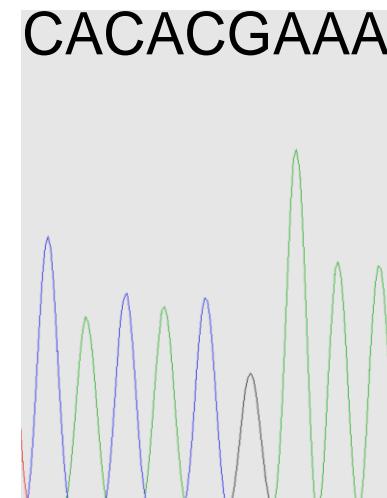
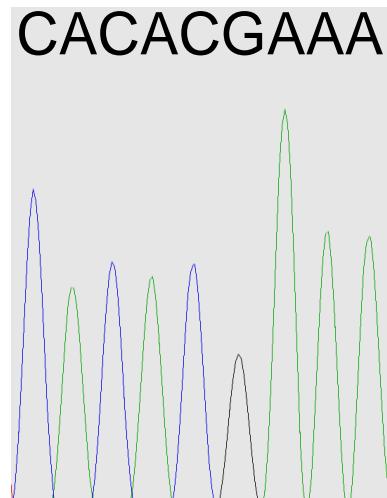
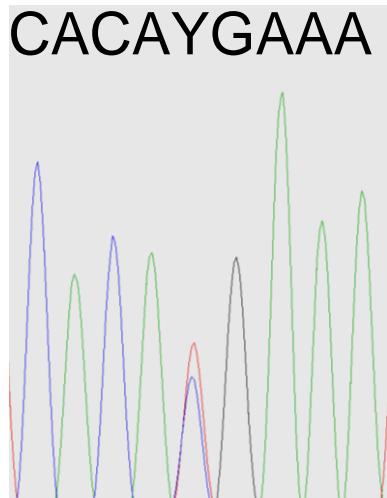


IC250E het

IC252E

IC253E

IC254E het



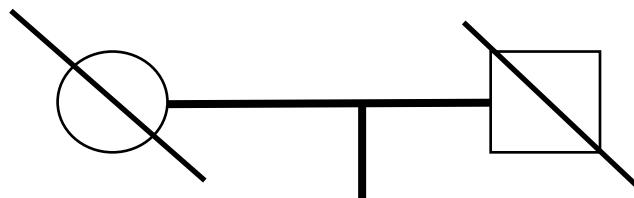
Deceased

○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

## ADCY10 Y402C

## Family F1



IC250E het

IC252E

IC253E

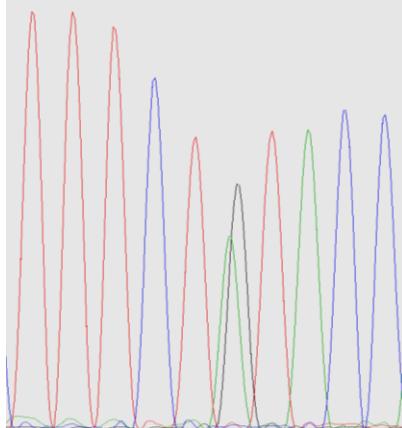
IC254E het

TTTCTRTACC

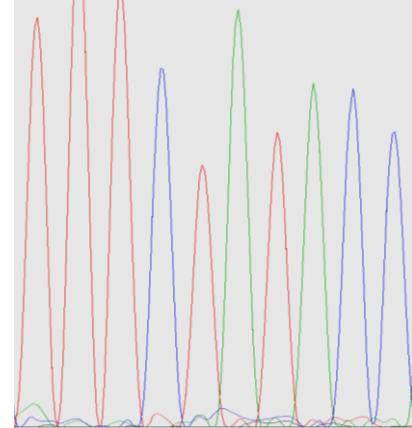
TTTCTATACC

TTTCTATACC

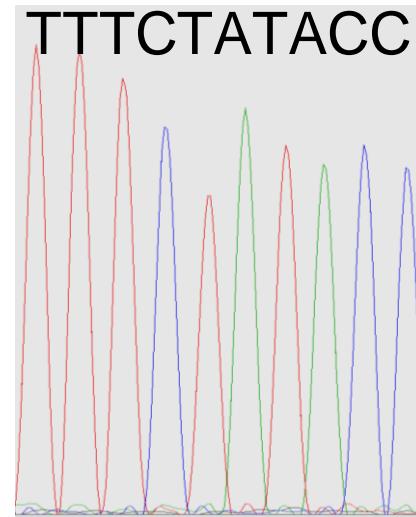
TTTCTRTACC



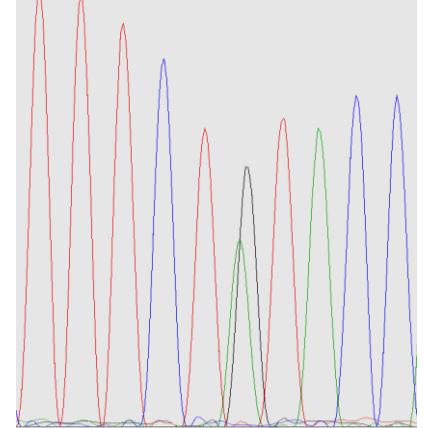
IC250E



IC252E



IC253E



IC254E

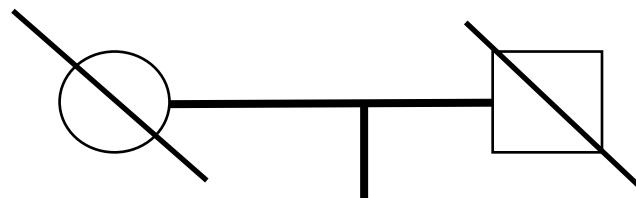
○ □ Deceased

○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

## MOCS1 Y402C

Family F1



IC250E het

IC252E

IC253E

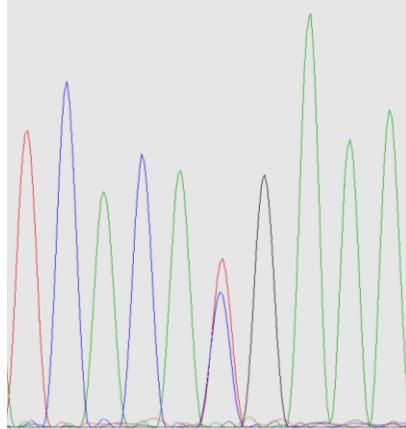
IC254E het

TCACACAYGCC

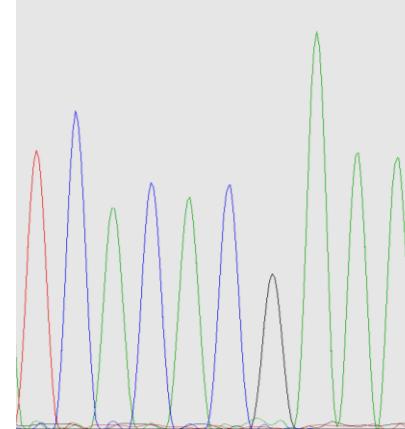
TCACACACGCC

TCACACACGCC

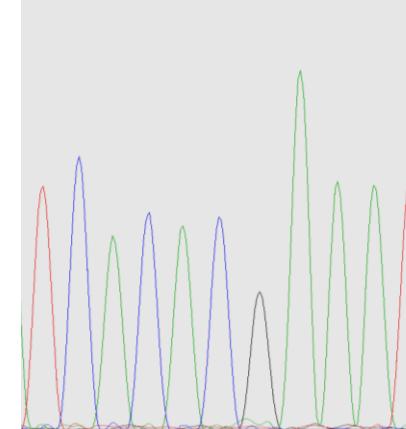
TCACACAYGCC



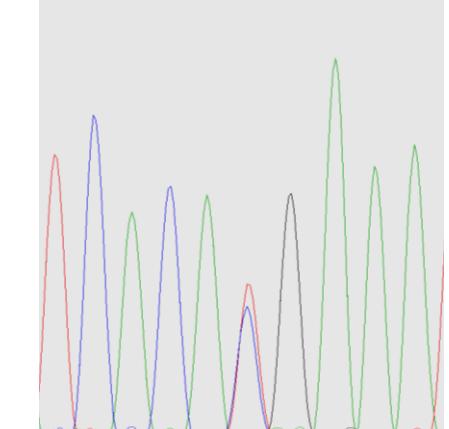
IC250E



IC252E



IC253E



IC254E

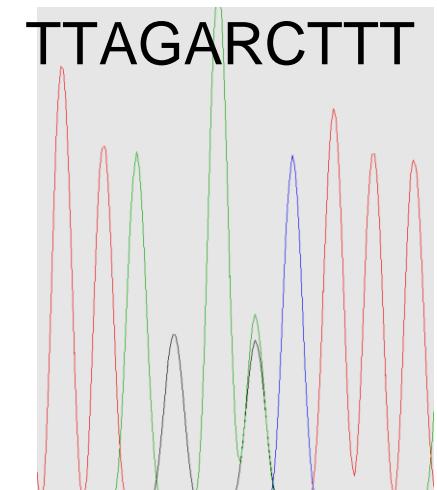
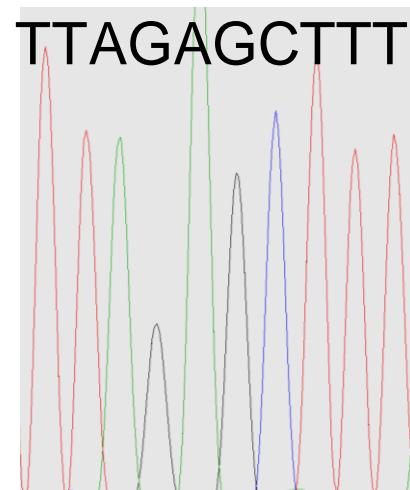
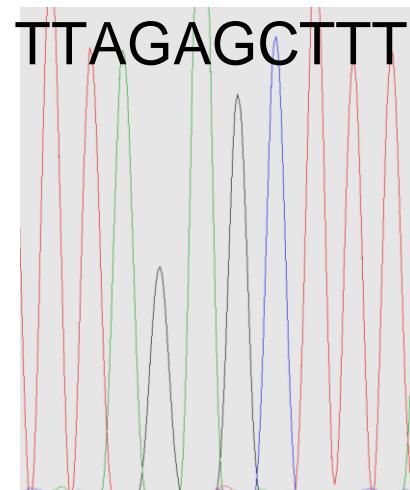
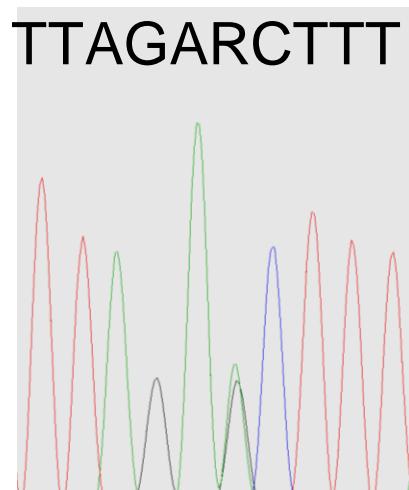
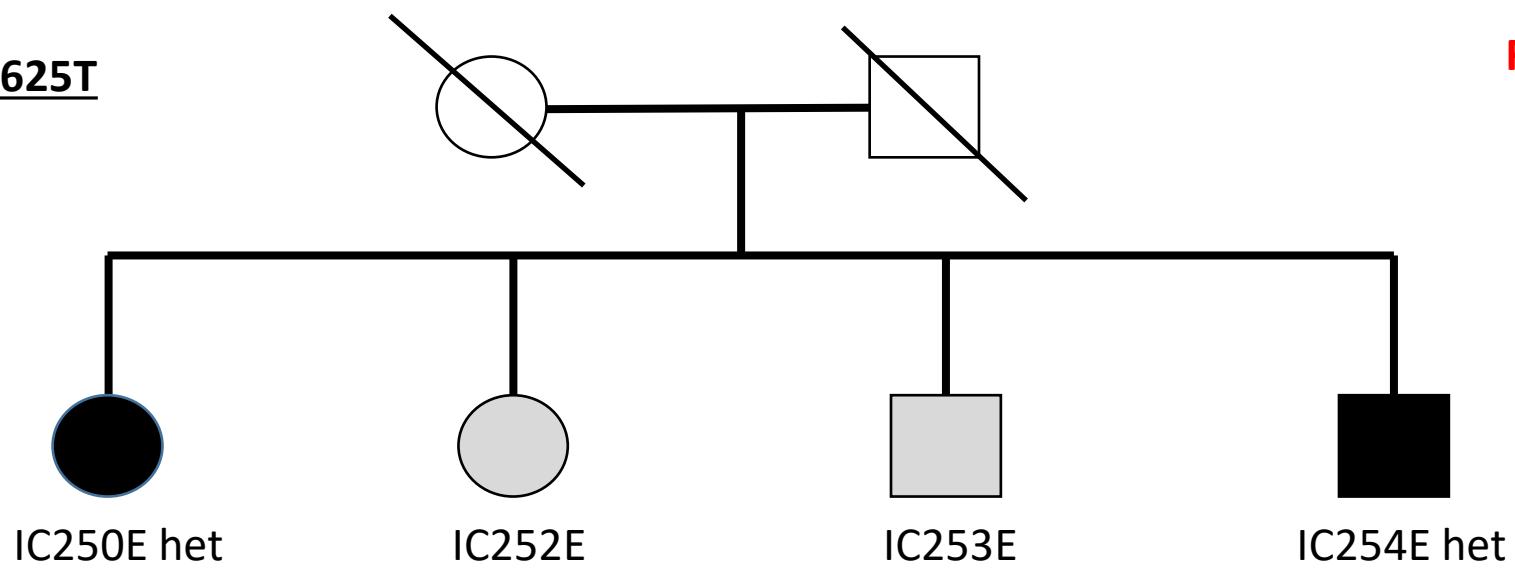
○ □ Deceased

○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

## ADGRG6 AD625T

Family F1



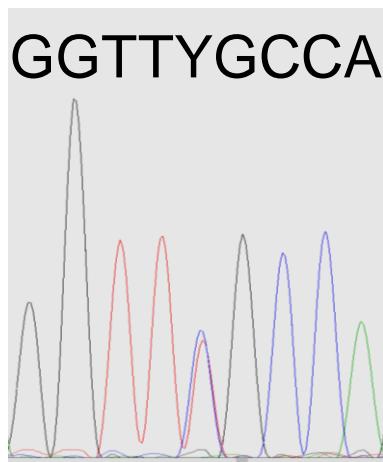
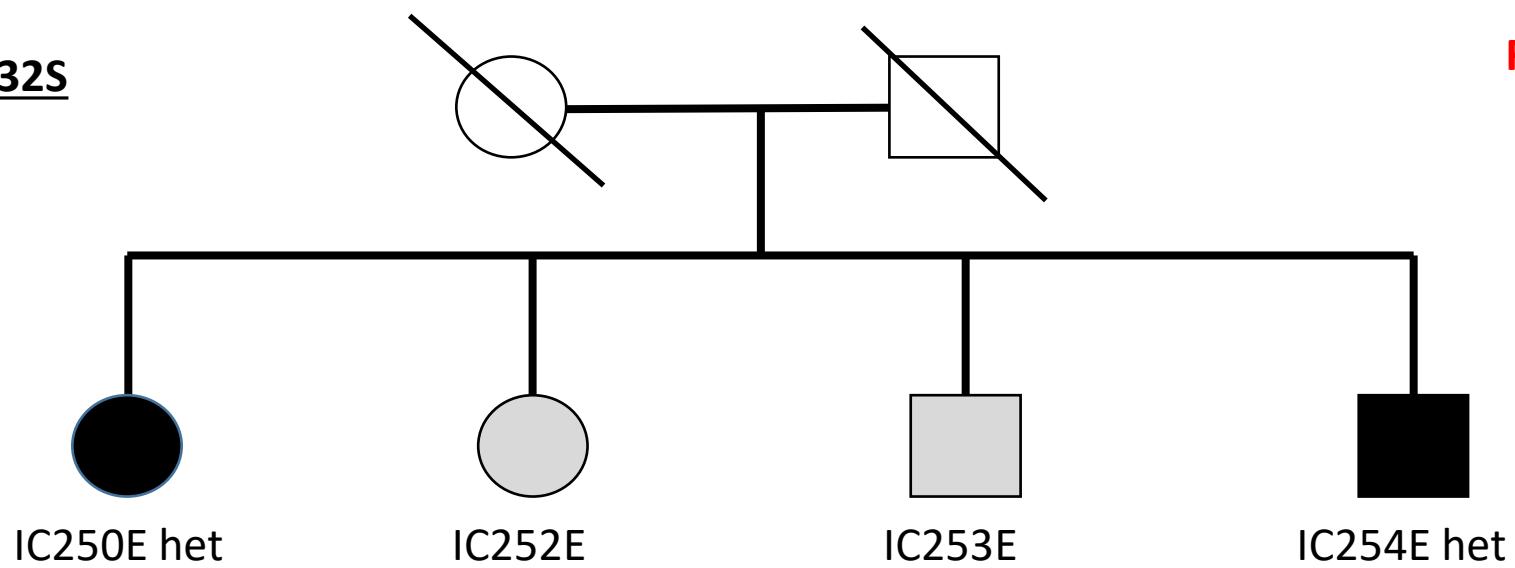
○ □ Deceased

○ □ Seropositive ASY patients

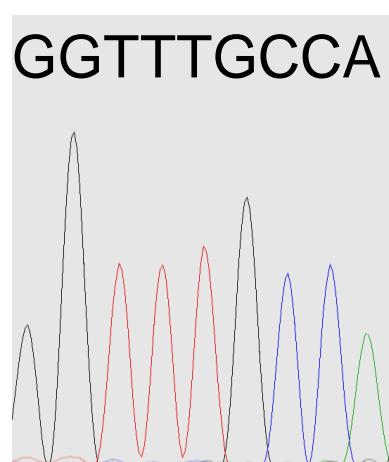
● ■ Seropositive CCC patients. het= heterozygote

## AKAP13 L1132S

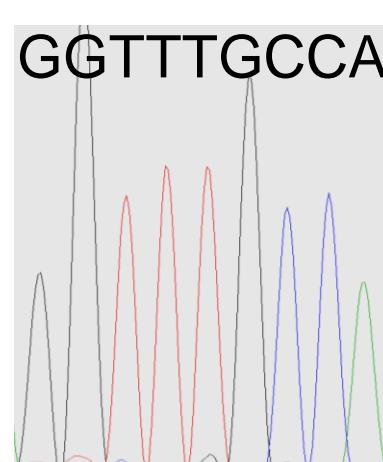
Family F1



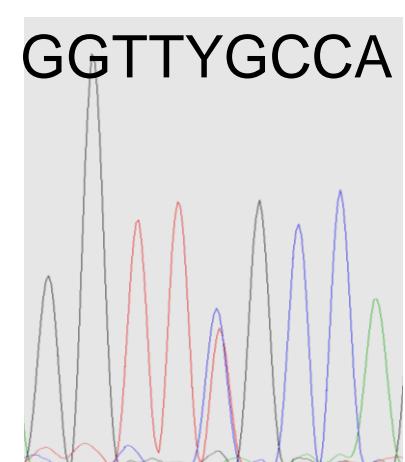
IC250E



IC252E



IC253E



IC254E

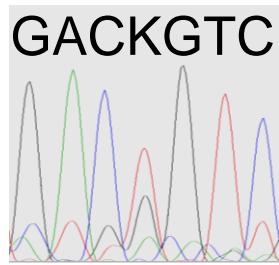
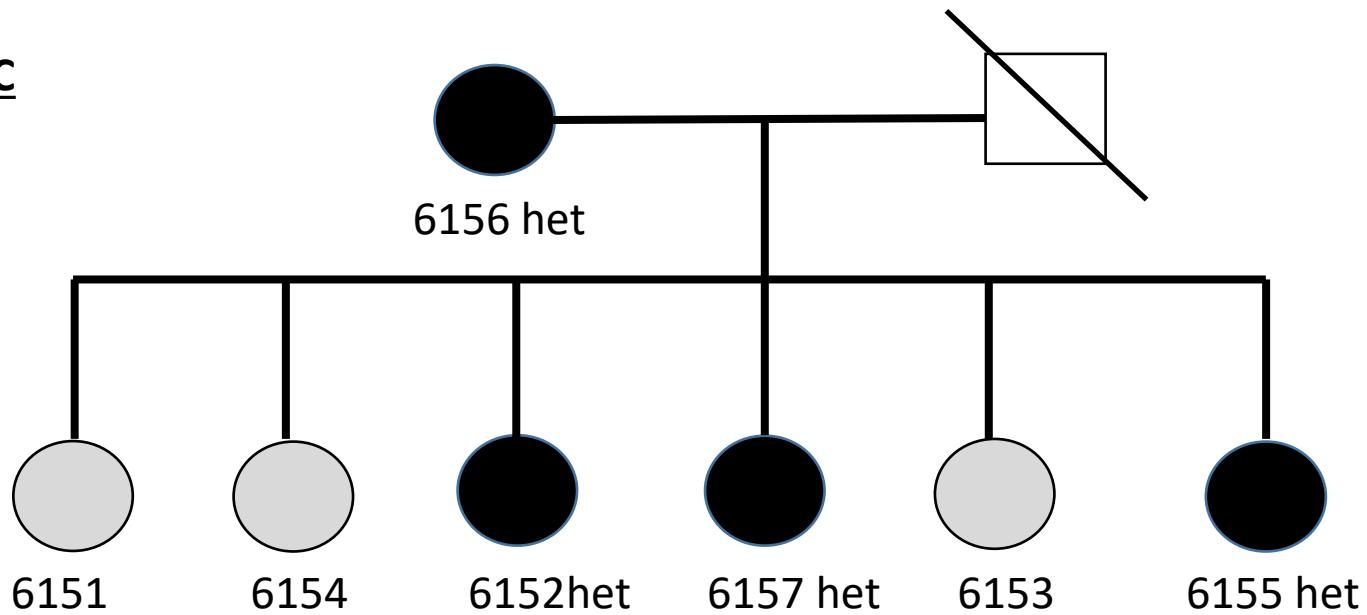
○ □ Deceased

○ □ Seropositive ASY patients

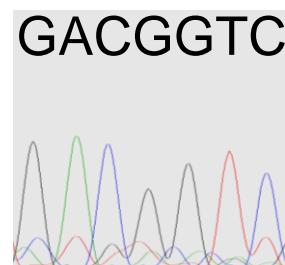
● ■ Seropositive CCC patients. het= heterozygote

## OBSCN G21113C

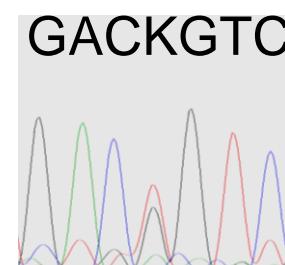
Family F2



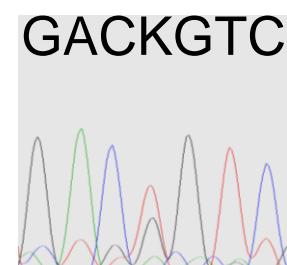
6156



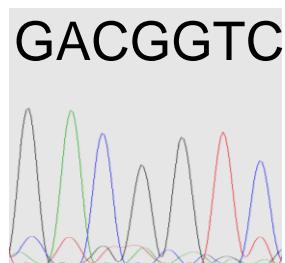
6154



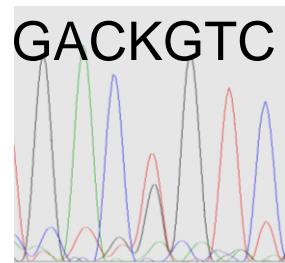
6157



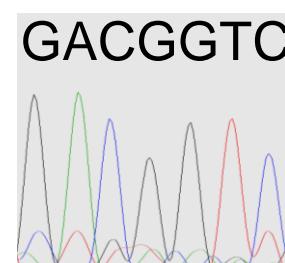
6155



6151



6152



6153

Deceased



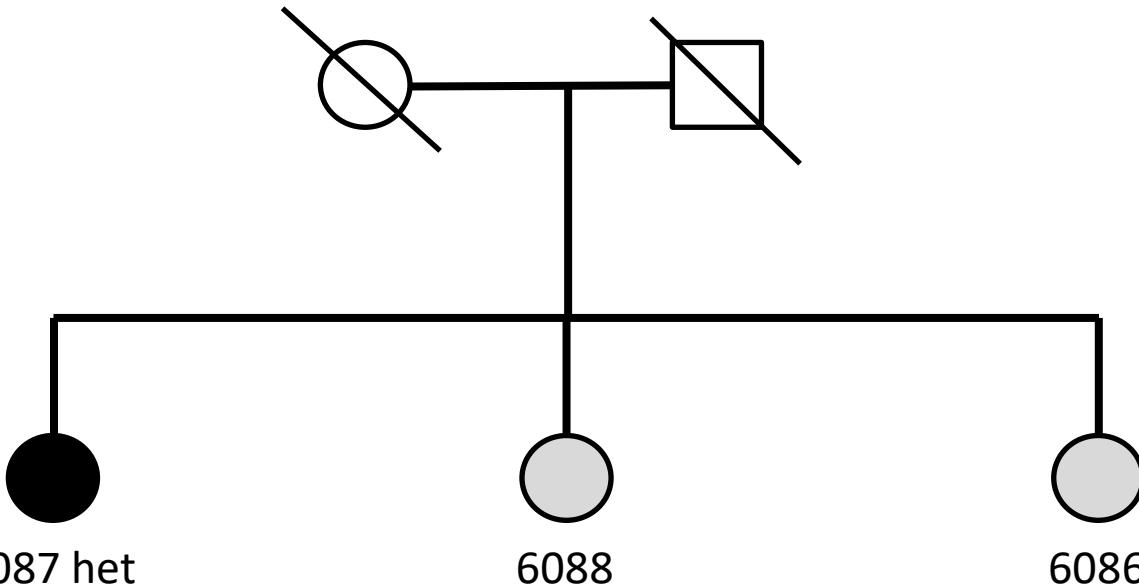
Seropositive ASY patients



Seropositive CCC patients. het= heterozygote

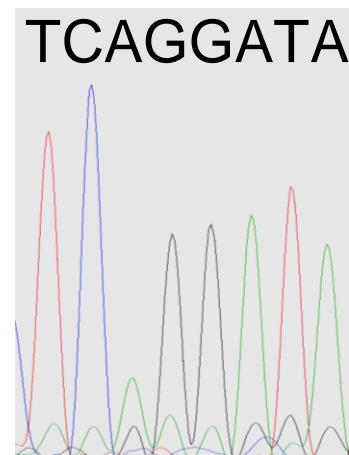
## APOB D749Y

Family F3



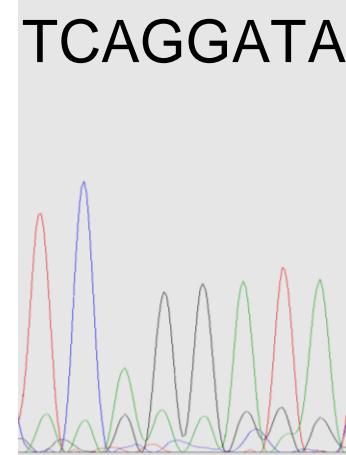
6087

Deceased



6088

Seropositive ASY patients

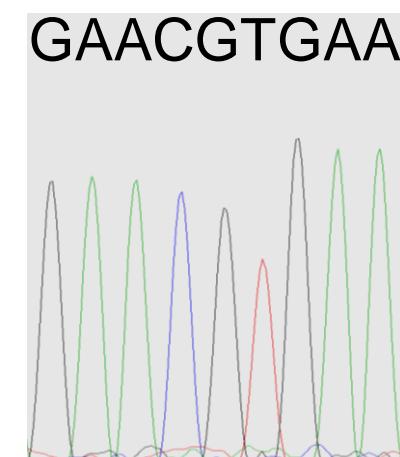
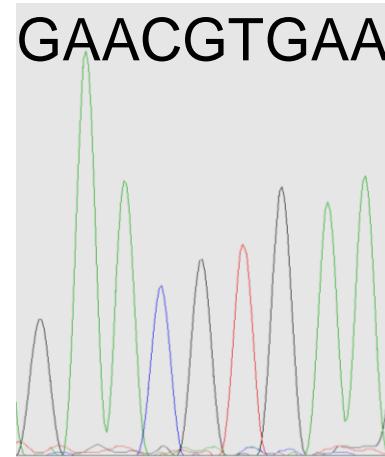
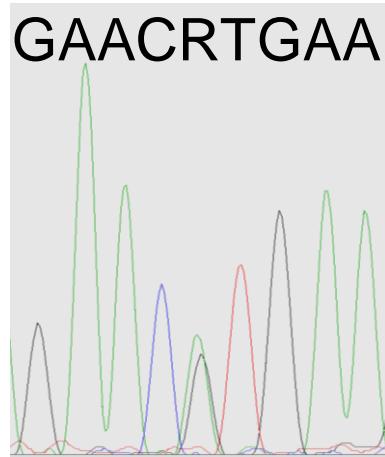
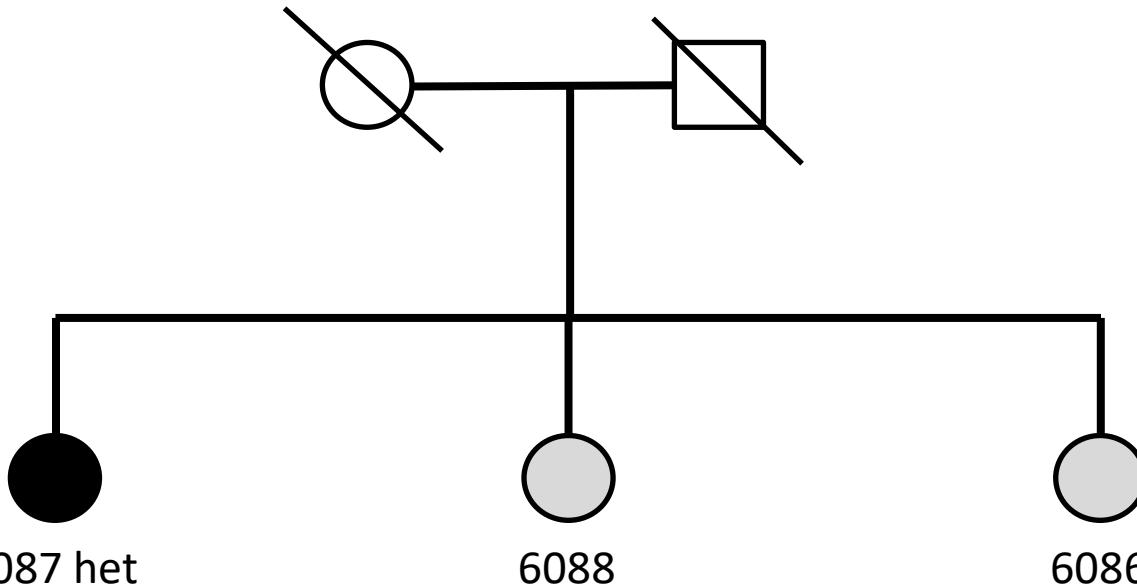


6086

Seropositive CCC patients. het= heterozygote

## MRPS18B V120M

Family F3



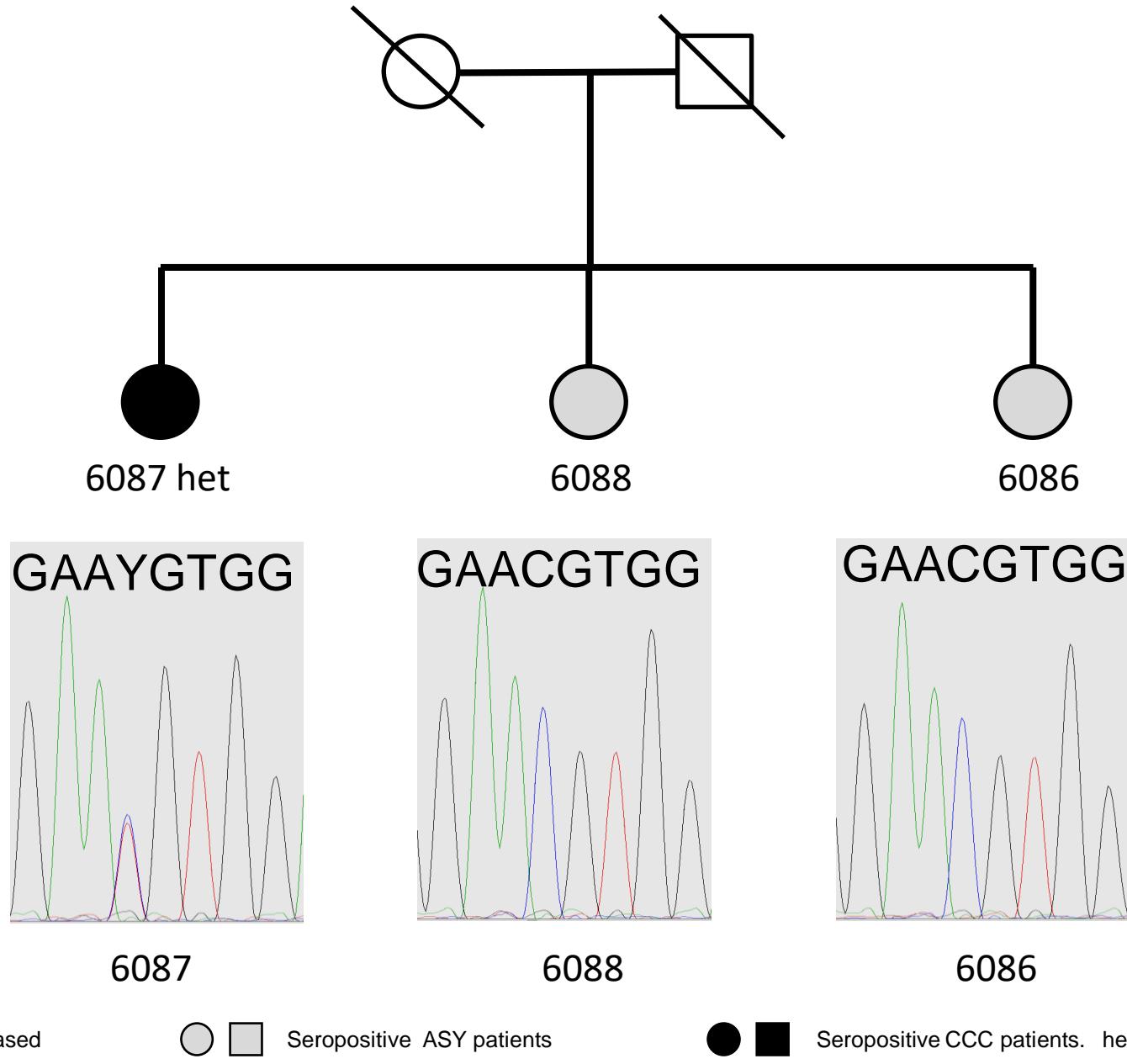
○ □ Deceased

○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

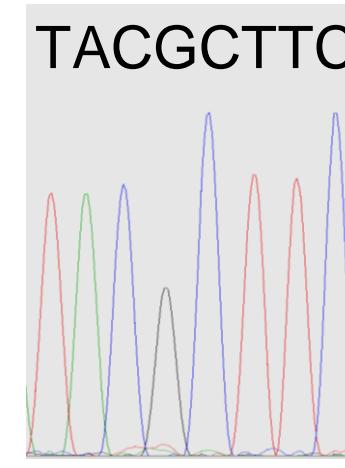
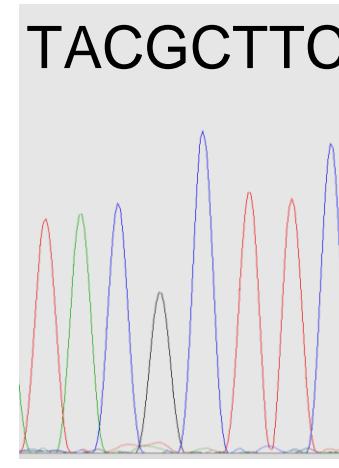
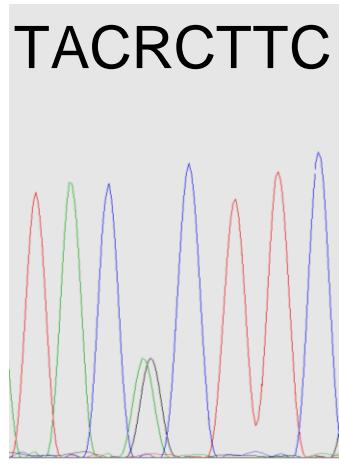
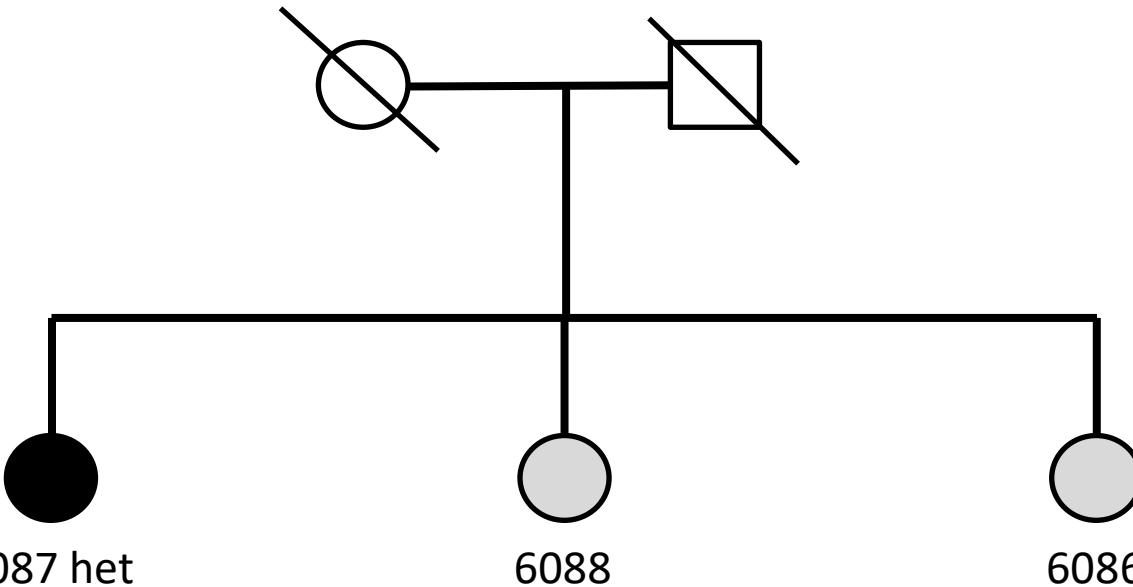
## PKHD1 T36M

## Family F3



## RNLS R222H

## Family F3



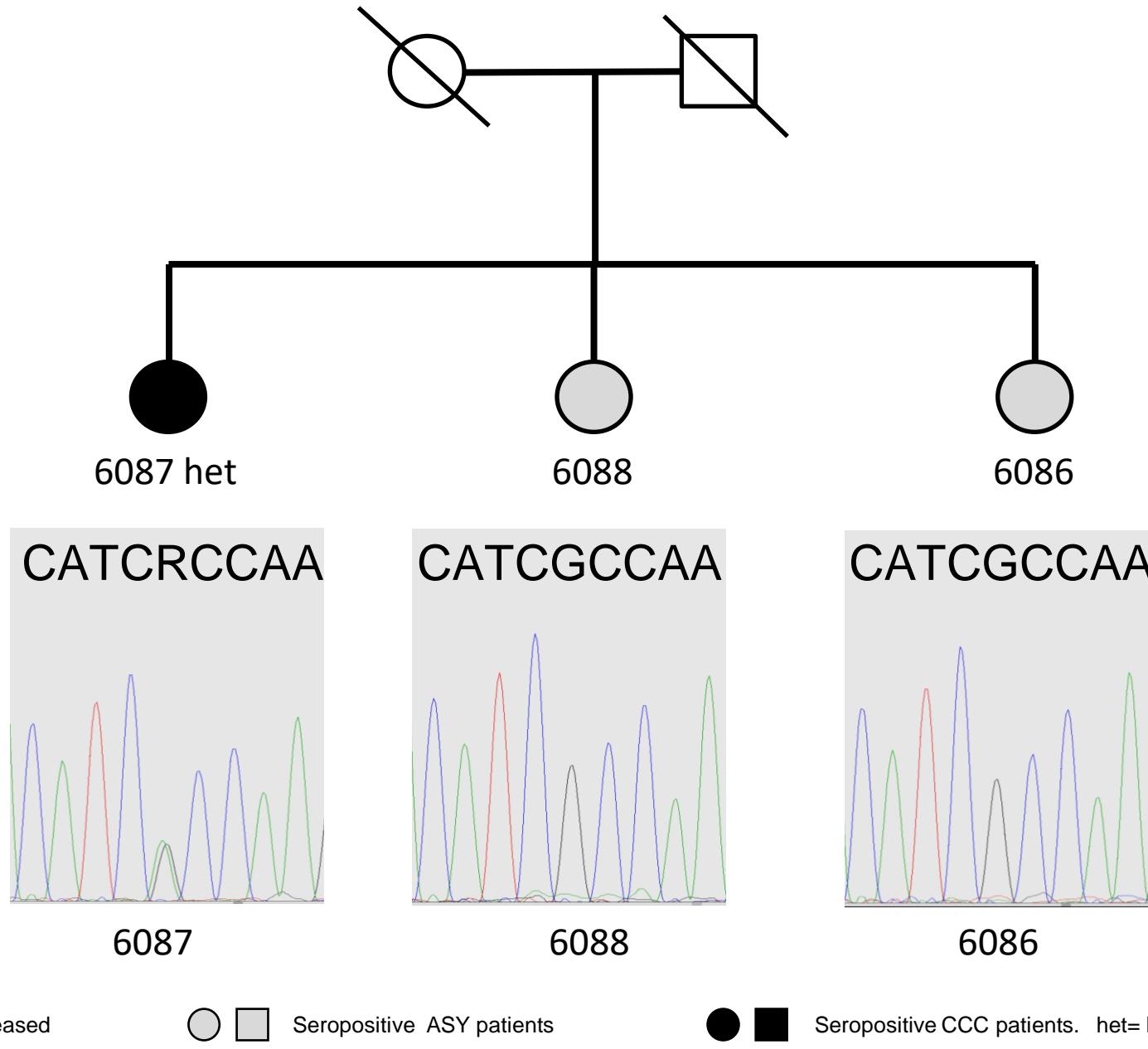
○ □ Deceased

○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

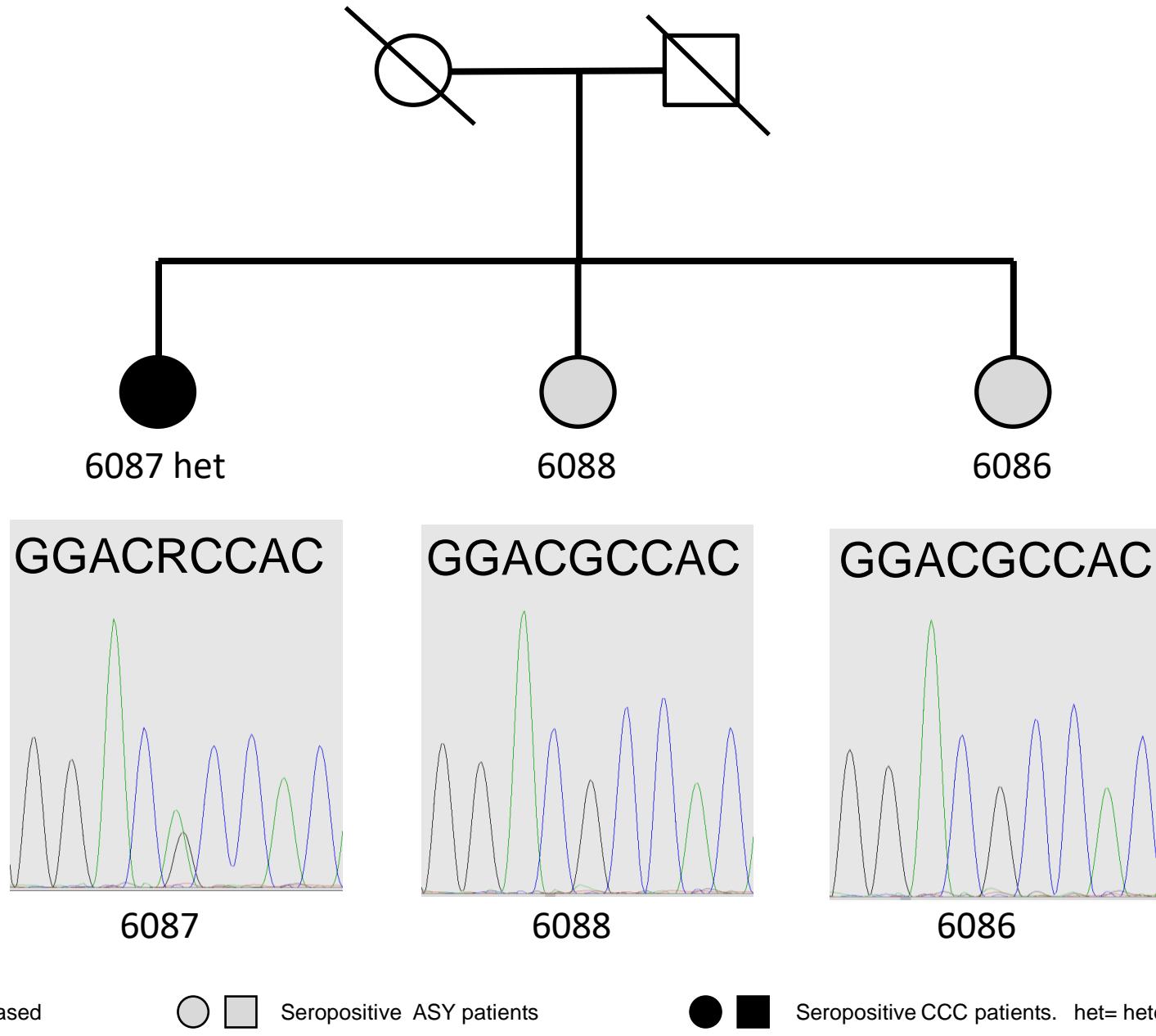
GIT1 A745T

Family F3



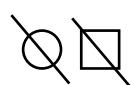
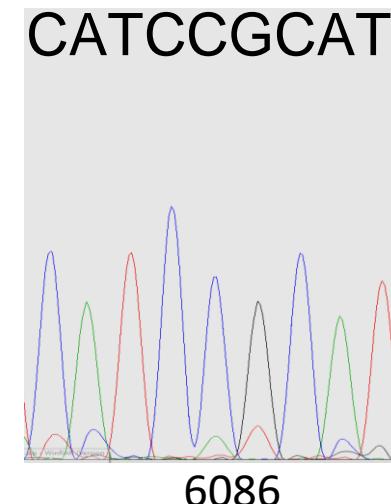
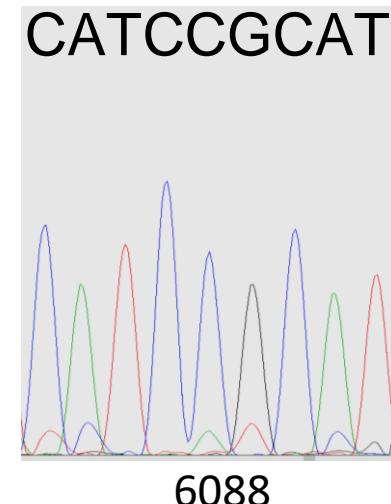
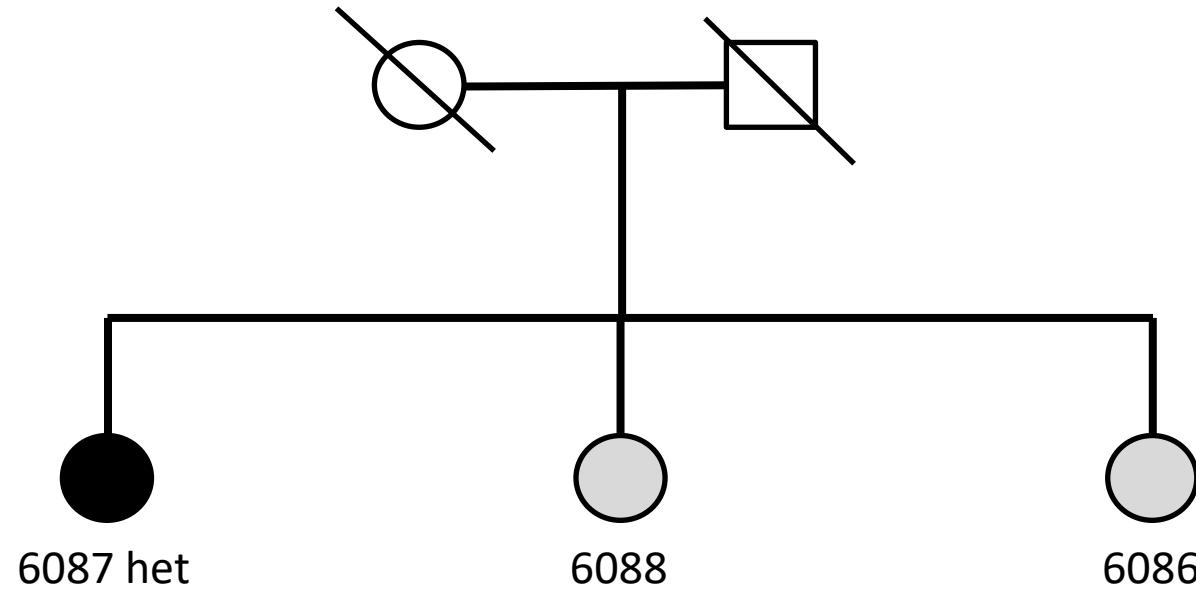
## GIT1 R43H

## Family F3



## LILRA2 R423C

## Family F3



Deceased



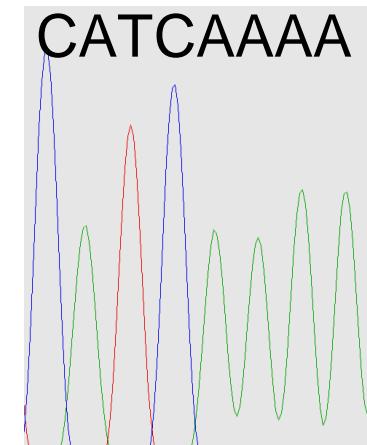
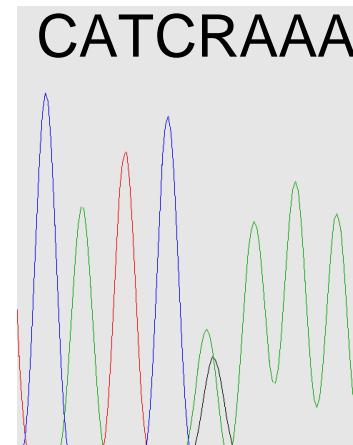
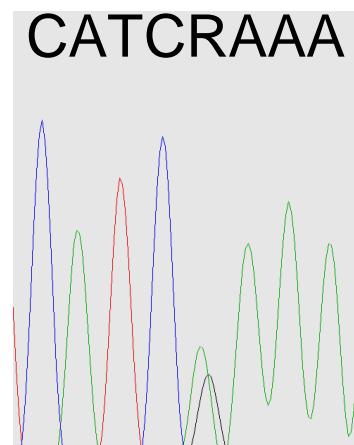
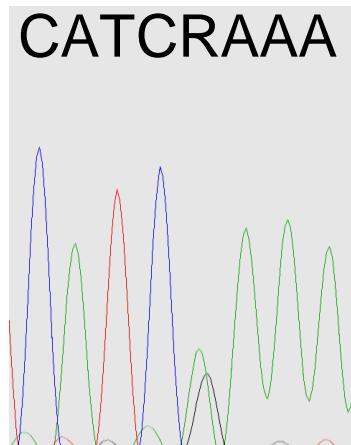
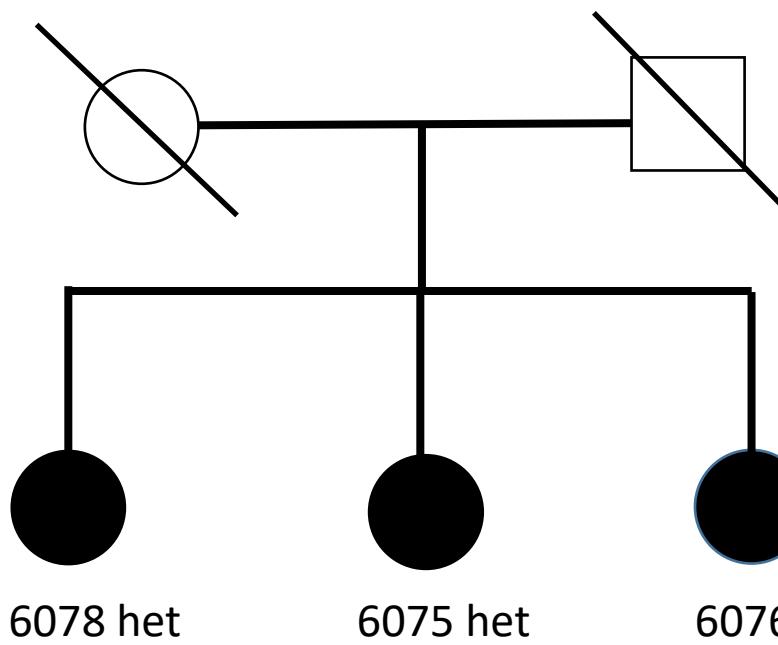
Seropositive ASY patients



Seropositive CCC patients

## MAP4K4 K91E

## Family F4

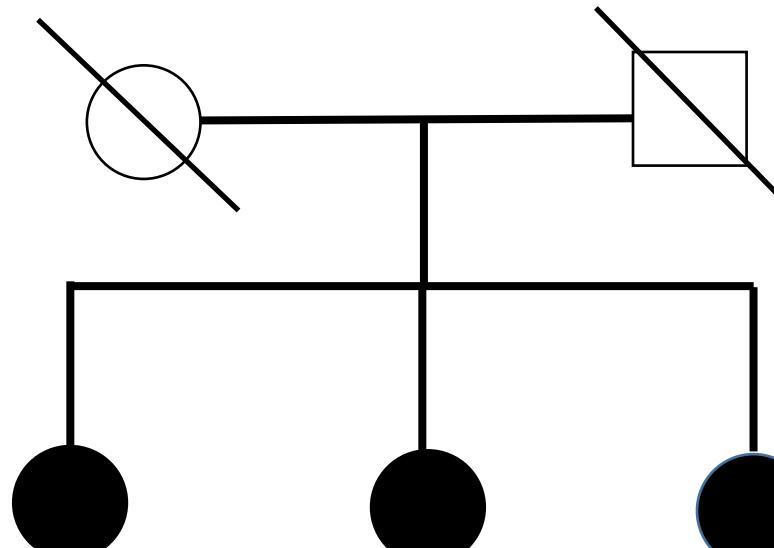


○ □ Deceased

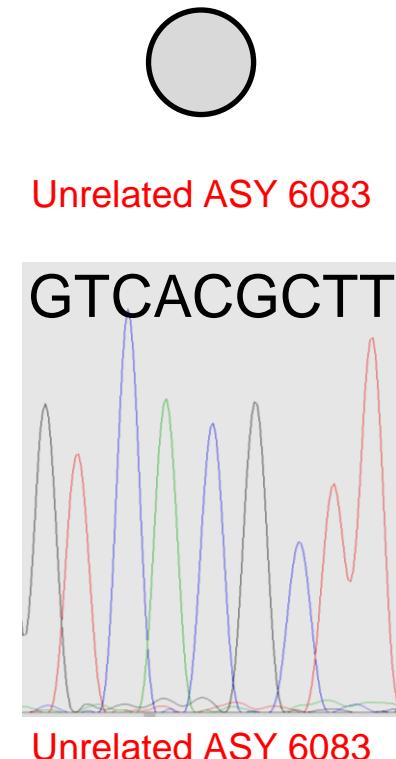
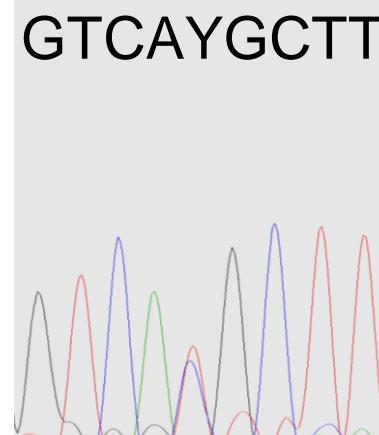
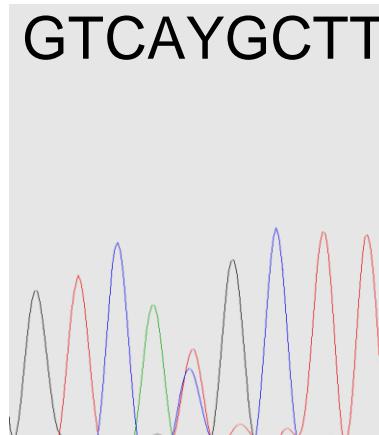
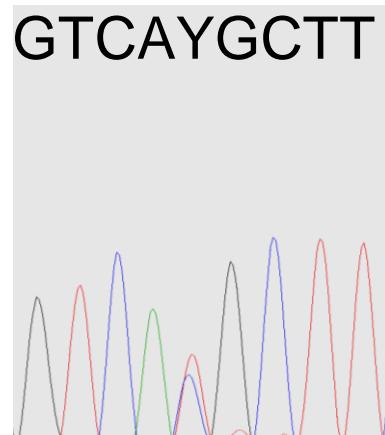
○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

### SLC11A1 R397C



Family F4

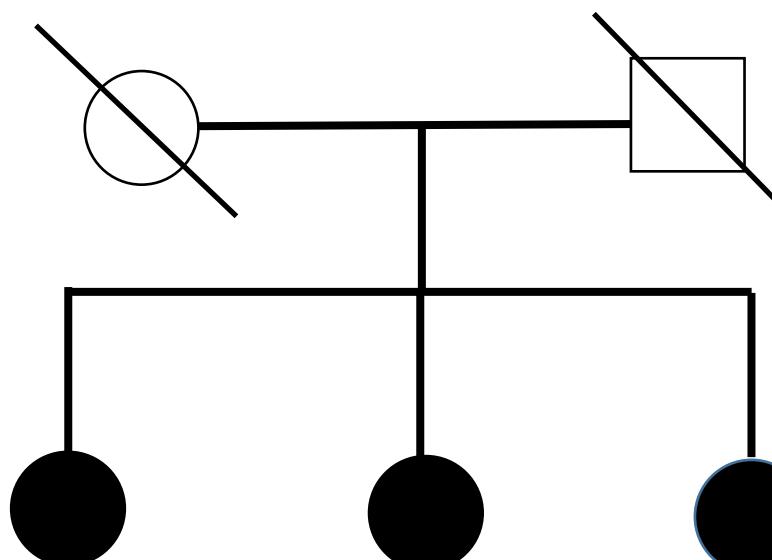


○ □ Deceased

○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

RPUSD3 W269X  
(stopgain)



Family F4

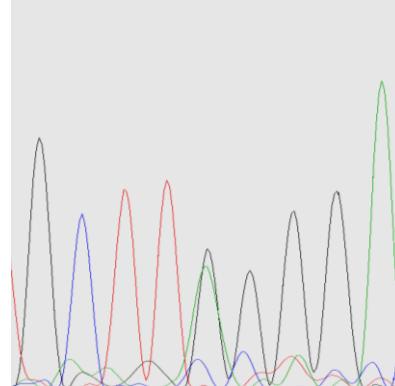
6078 het

6075 het

6076 het

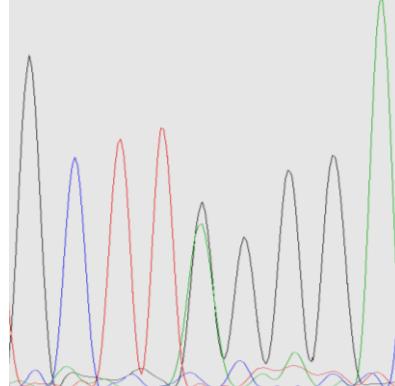
Unrelated ASY 6083

GCTTRGGGA



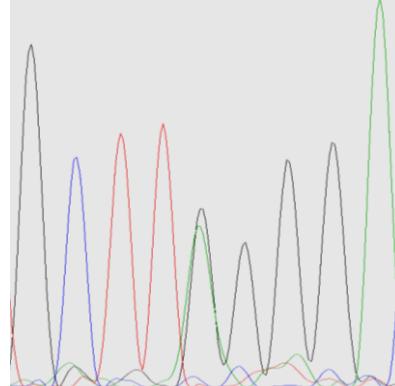
6078

GCTTRGGGA



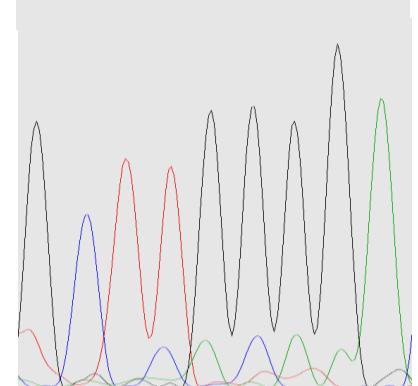
6075

GCTTRGGGA



6076

GCTTGGGGA



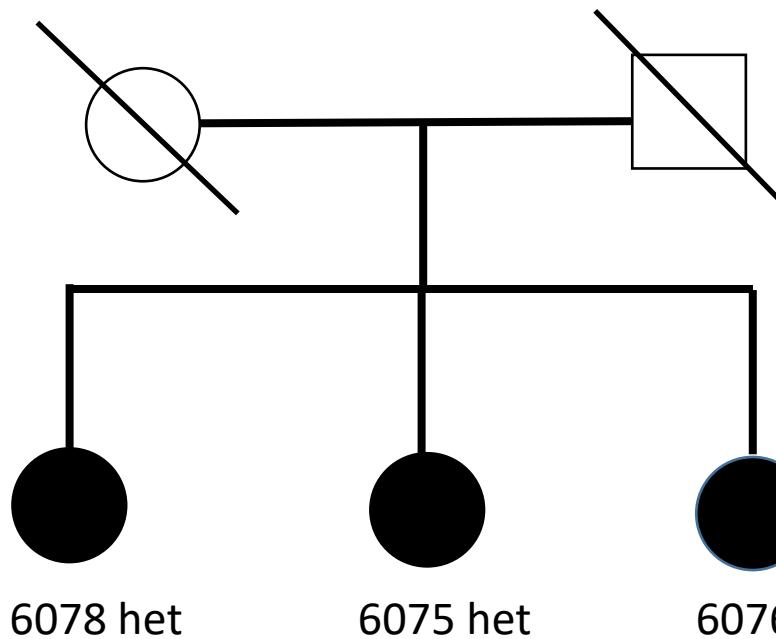
Unrelated ASY 6083

Deceased

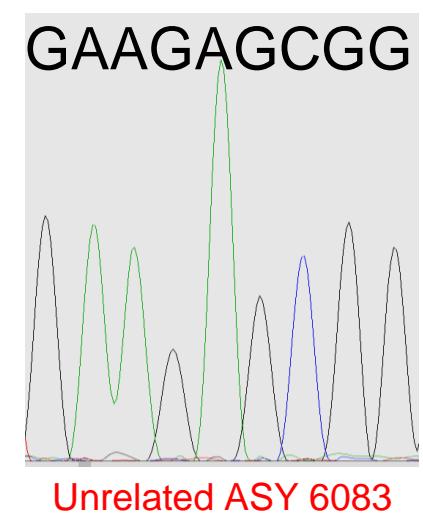
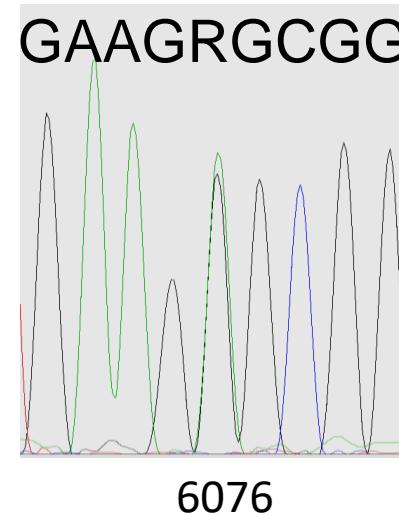
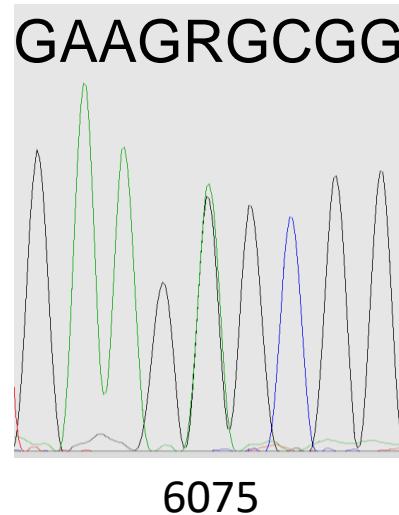
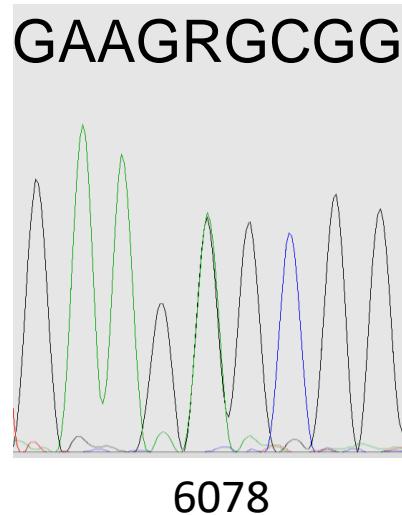
Seropositive ASY patients

Seropositive CCC patients. het= heterozygote

## UMPS S30G



Family F4



○ □ Deceased

○ □ Seropositive ASY patients

● ■ Seropositive CCC patients. het= heterozygote

## Family F5

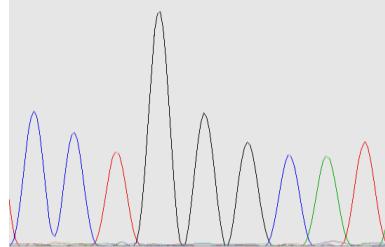
### MAML1 G136E



6136 het

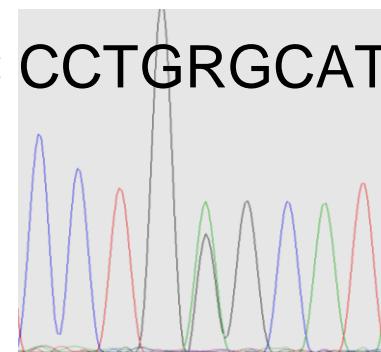
6131 het

CCTGGGCAT



6135 het

CCTGRGCAT



6134 het

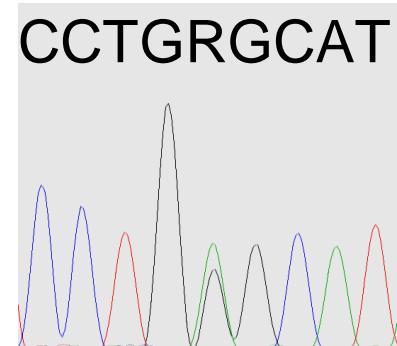
6131

6133

6136

CCTGRGCAT

CCTGRGCAT



6135

6134



Deceased



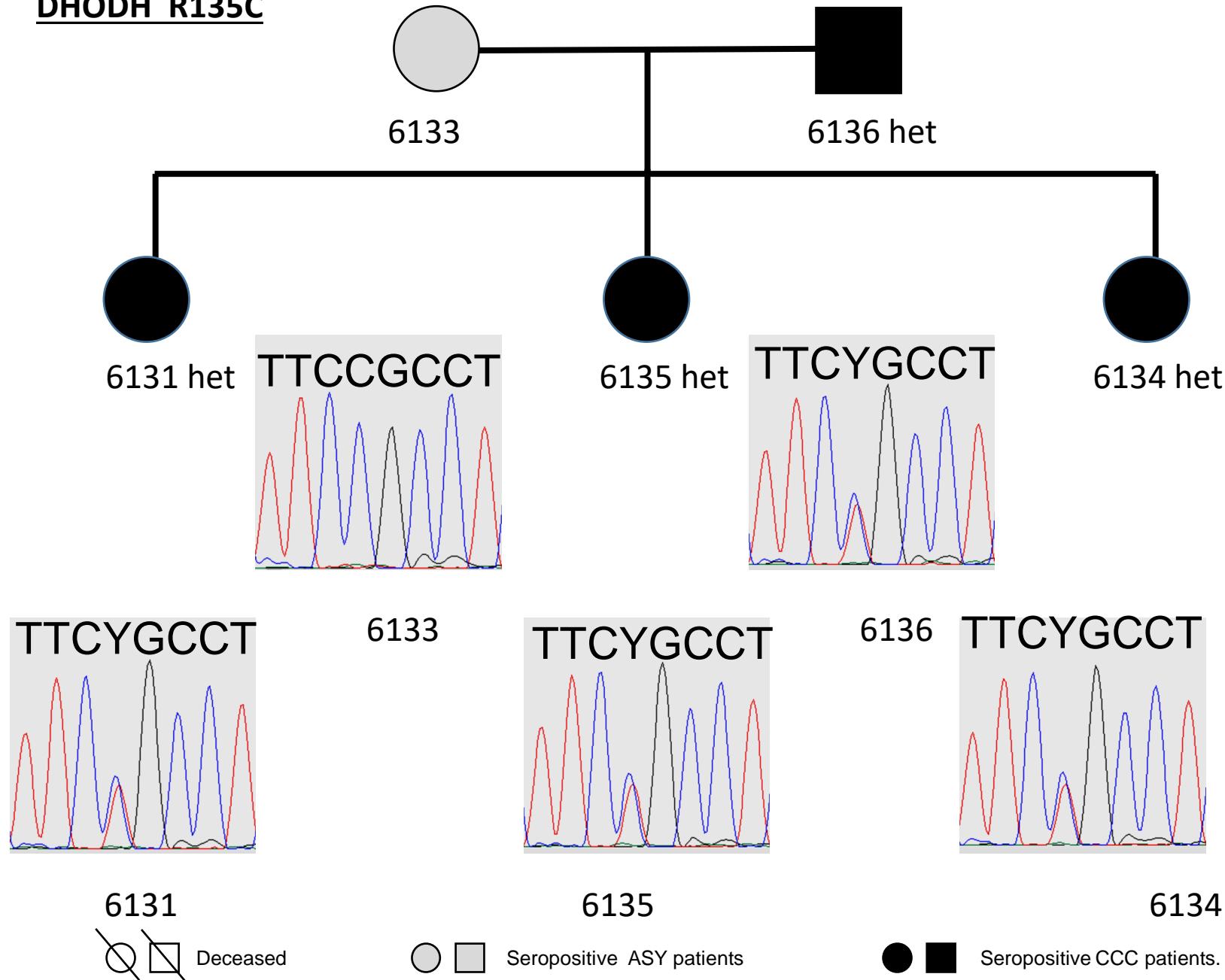
Seropositive ASY patients



Seropositive CCC patients. het= heterozygote

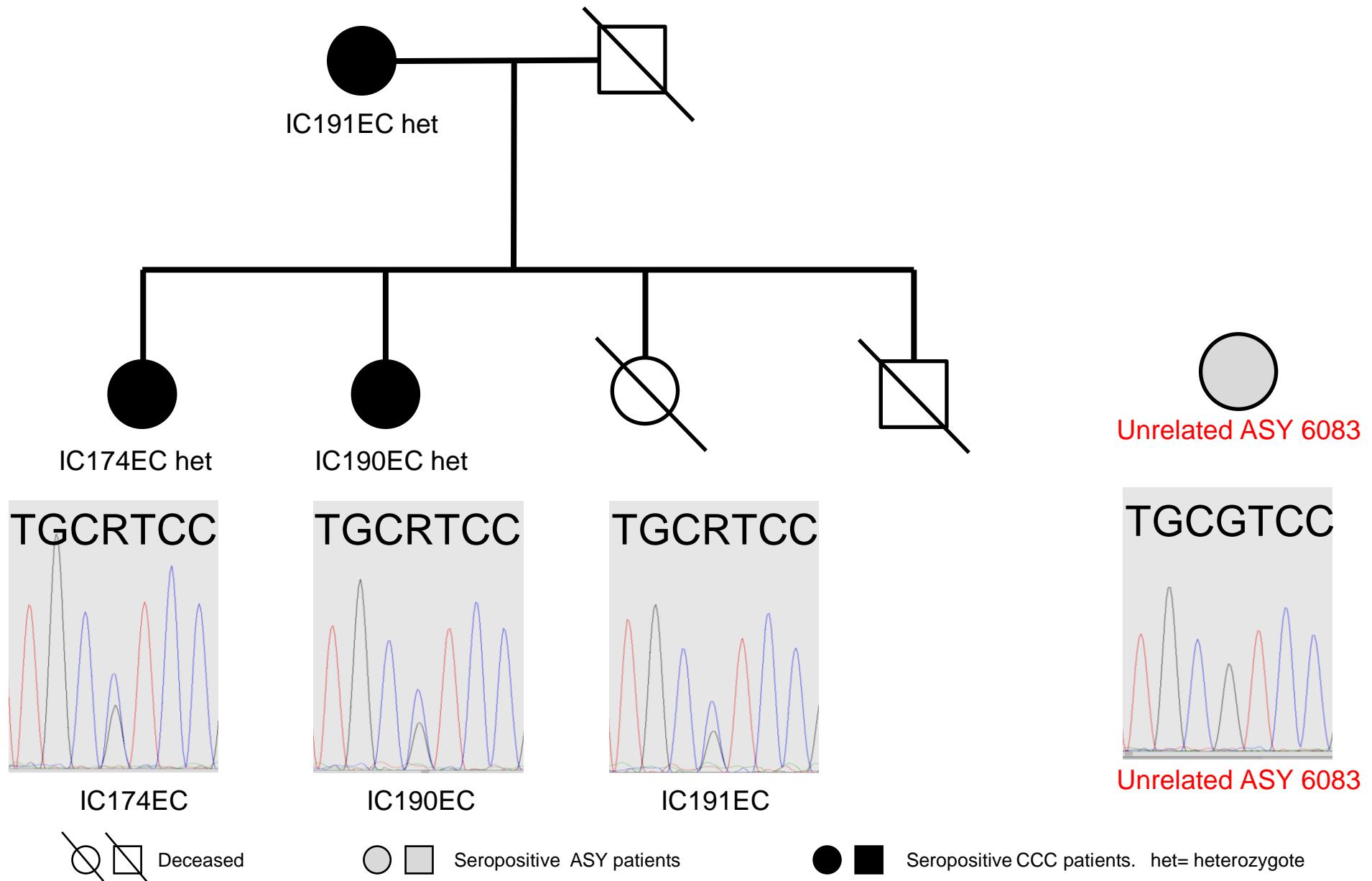
## Family F5

### DHODH R135C



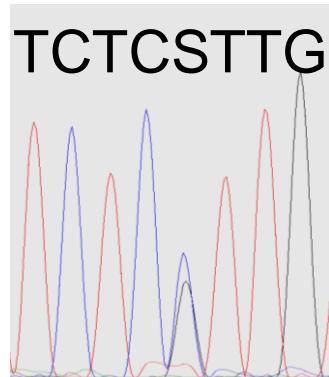
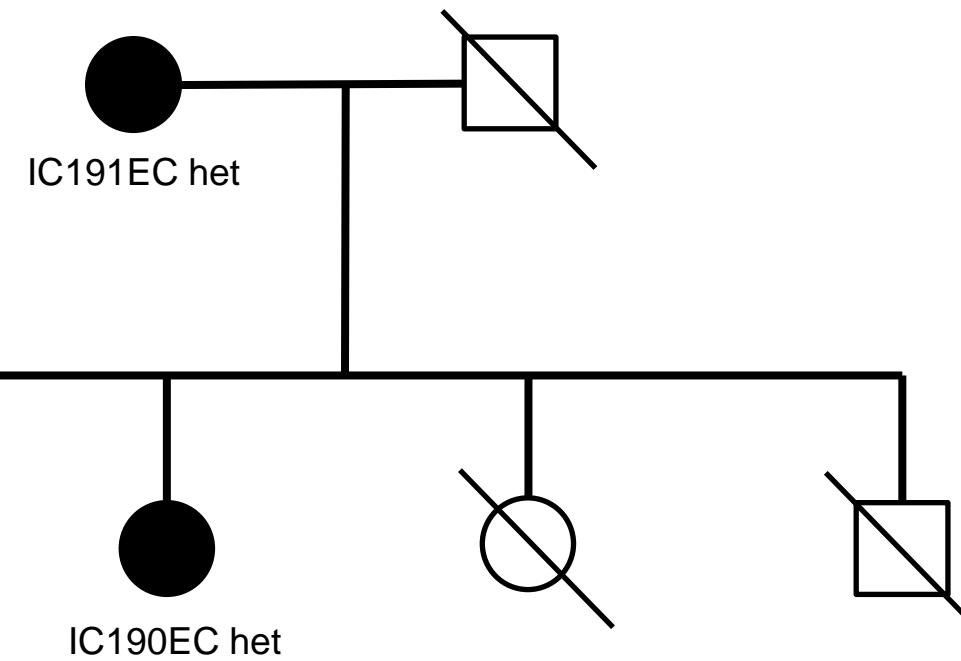
## TNFRSF4 D163E

Family F6

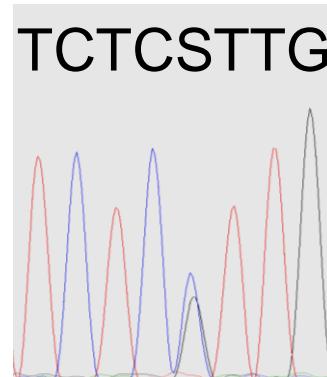


## APOB N3107K

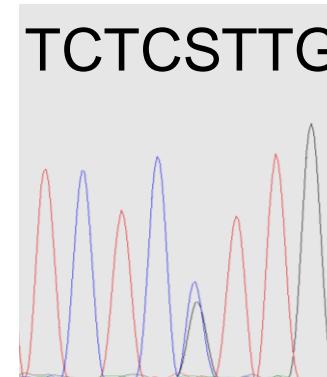
Family F6



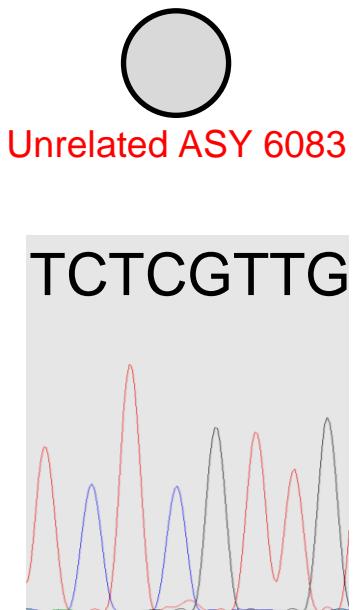
IC174EC



IC190EC



IC191EC



Unrelated ASY 6083

Deceased



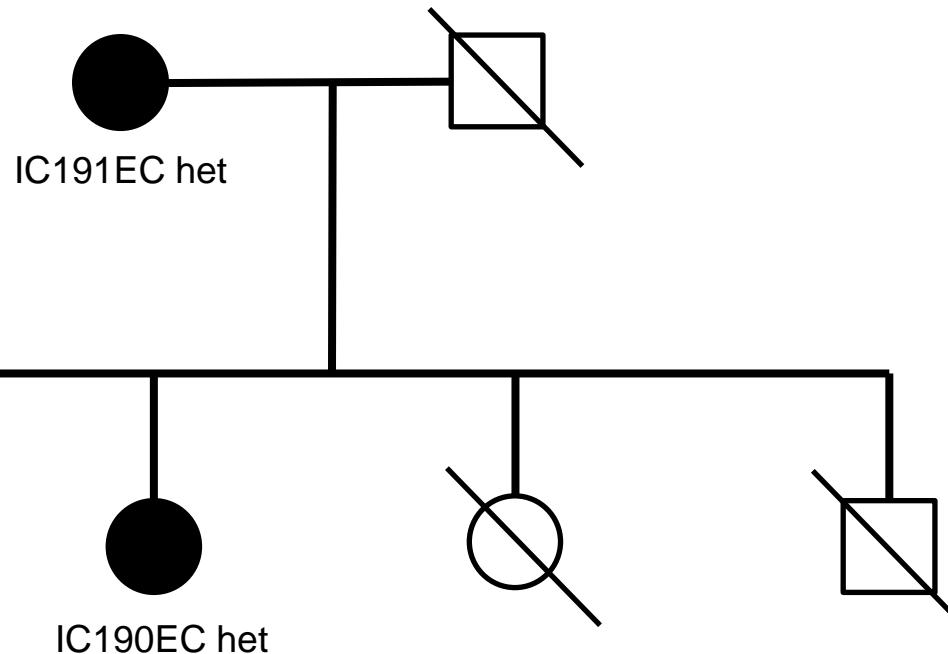
Seropositive ASY patients



Seropositive CCC patients. het= heterozygote

## SERPINE2 M64T

Family F6



IC174EC het

IC190EC het

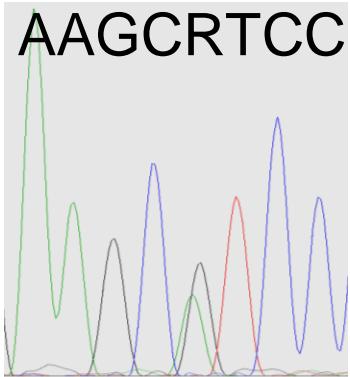
Unrelated ASY 6083

AAGCRTCC

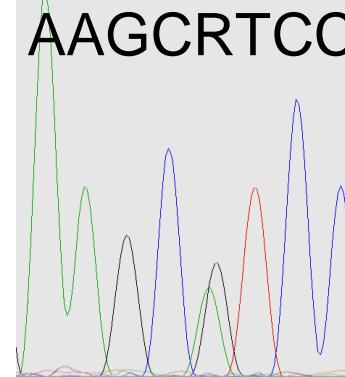
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AAGCRTCC

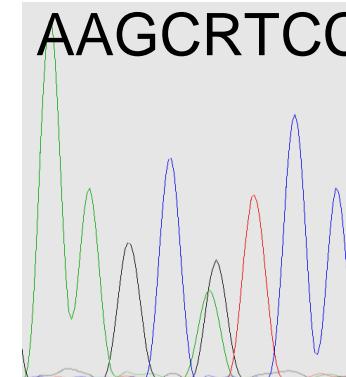
AAGCATCC



IC174EC



IC190EC



IC191EC

Unrelated ASY 6083

Deceased

Seropositive ASY patients



Seropositive CCC patients. het= heterozygote